

BILE SALTS, profile (urine and plasma)

Relevant disorders

Bile salt analysis is an essential tool in the diagnosis of disorders of bile acid biosynthesis:

1. 3β -hydroxysteroid dehydrogenase deficiency;
2. 3-oxo- Δ^4 steroid reductase deficiency;
3. C27-hydroxylase deficiency (Cerebrotendinous Xanthomatosis);
4. 7 α -hydroxylase (oxosterol deficiency) ;
5. Peroxisomal disorders where β -oxidation of C27 bile acids to primary (C24) bile acids is defective.
6. C25-hydroxylase deficiency.
7. Bile acid conjugation deficiency.

Detection of increased primary bile salts compared to controls can also reveal cholestatic liver disease.

N.B. This is for the diagnosis of bile acid synthesis disorders (NOT the same as total bile acid/bile salts quantitation i.e. **not for cholestasis in pregnancy**)

Related Metabolic Tests

Very long chain fatty acids
Plasmalogens
Sterols (cholestanol)

Methodology

Negative ion electrospray tandem mass spectrometry using a deuterated internal standard (D4 taurocholate) for quantitation.

Sample requirements

5ml random urine sample, in a plain universal tube.
1ml lithium heparin plasma or serum.

Turn Around Time

6 weeks

Transport information/Contact details

Send by first class post to:

Department of Clinical Chemistry
Sheffield Children's NHS Foundation Trust
Western Bank, Sheffield
S10 2TH, UK

Jane Dalley (Clinical Scientist)
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Reference Ranges

N.B: these reference ranges are only valid with data produced by the Department of Clinical Chemistry, Sheffield Children's NHS Foundation Trust.

	Plasma $\mu\text{mol/L}$	Urine $\mu\text{mol}/\text{mmol creat.}$
Glycodihydroxycholanoate	< 6	0.02 – 0.47
Glycotrihydroxycholanoate	< 2	0.04 – 1.39
Taurodihydroxycholanoate	< 2	0.01 – 0.02
Tauotrihydroxycholanaote	< 2	0.01 – 0.08

Interpretation will be provided as part of the report.

References

- Mills et al. A method for the Quantitation of Conjugated Bile Acids in Dried Blood Spots Using Electrospray Ionisation-Mass Spectrometry. Pediatric Research (1998) 43 No.3 p361-367
- Bootsma et al. Rapid analysis of conjugated bile acids in plasma using electrospray tandem mass spectrometry: Application for selective screening of peroxisomal disorders. J. Inher. Metab. Dis. 22 (1999) p307-310
- K. Bove Liver disease caused by disorders of bile acid synthesis. Clinics in Liver Disease Vol 4 No. 4 November 2000.